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Research Letter

Prenatal diagnosis of mosaicism for trisomy 2 in a single colony at amniocentesis in a pregnancy with a favorable outcome

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Dear Editor,

A 36-year-old, gravida 3, para 1, woman underwent amniocentesis at 18 weeks of gestation because of advanced maternal age. Cytogenetic analysis of the cultured amniocytes revealed a karyotype of 47,XY,+2[1]/46,XY[21]. Among 22 colonies of cultured amniocytes, one colony had three cells, and all cells of this colony had the karyotype of 47,XY,+2, whereas the other 21 colonies had the karyotype of 46,XY. Level II ultrasound findings were unremarkable. Repeat amniocentesis was performed at 23 weeks of gestation. Interphase fluorescence *in situ* hybridization (FISH), array comparative genomic hybridization (aCGH) and quantitative fluorescent polymerase chain reaction (QF-PCR) were applied on uncultured amniocytes, and conventional cytogenetic analysis was applied on cultured amniocytes. The parental karyotypes were normal. QF-PCR analysis excluded uniparental disomy 2. aCGH using Roche ISCA Plus Cytogenetic Array (Roche, Madison, WI, USA) revealed no genomic imbalance. Interphase FISH analysis on uncultured amniocytes using the bacterial artificial chromosome probe of RP11-332H14 [2q12.1, fluorescein isothiocyanate (FITC)] revealed trisomy 2 in 6/112 (5.3%) cells comparing with 1/52 (1.9%) cell in the normal control. Cytogenetic analysis of the cultured amniocytes revealed a karyotype of 46,XY in all 20 colonies examined. The parents decided to continue the pregnancy. At 38 weeks of gestation, a 2906-g healthy male infant was delivered with no phenotypic abnormalities. Cytogenetic analysis of cord blood revealed a karyotype of 46,XY in 40/40 lymphocytes examined. The neonate was phenotypically normal and had normal psychomotor development at age two years during postnatal follow-ups.

We previously reported prenatal diagnosis of mosaicism involving trisomy 2 at amniocentesis with a favorable outcome [1–3]. In this presentation, we report an additional case of prenatal diagnosis mosaicism for trisomy 2 in a single colony associated with a favorable outcome. However, prenatal diagnosis of mosaic trisomy 2 at amniocentesis should alert abnormalities on fetal ultrasound such as intrauterine growth restriction (IUGR), oligohydramnios, single umbilical artery, congenital heart defects, facial dysmorphism, cleft palate and lip, neural tube defects, diaphragmatic hernia, ventriculomegaly and hydronephrosis [1,4–6]. Chen et al. [5] reported prenatal diagnosis of trisomy 2 in 26.7% (8/30 colonies) of cultured amniocytes and in 11.1% (13/117 cells) of uncultured amniocytes in a pregnancy associated with severe oligohydramnios, IUGR, ventricular septal defect, preaxial polydactyly and facial dysmorphism in the fetus. Tuğ et al. [6] reported prenatal diagnosis of trisomy 2 in 14% (12/85 cells) of cultured amniocytes in a pregnancy associated with single umbilical artery, cardiac dextroposition and diaphragmatic hernia in the fetus. Prenatal diagnosis of mosaic trisomy 2 at amniocentesis in pregnancy associated with severe IUGR should also raise a suspicion of coexistence of complete trisomy 2 in the placenta [4,5].

Conflict of interest

The authors have no conflicts of interest relevant to this article.

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